

Partial Biotinidase Deficiency

General Overview

Q. What is biotinidase deficiency?

A. Biotinidase deficiency is a treatable disorder that affects the way the body recycles biotin, one of the B12 vitamins. Because this system doesn't work properly, the body has low levels of biotin. This vitamin is necessary for many functions in the body.

Q. Is there only one form of biotinidase deficiency?

A. No. Partial biotinidase deficiency is a less severe form.

Q. How does the body normally process biotin?

A. The body gets biotin from the food we eat and conserves it by recycling. Biotin attaches to proteins during metabolism. Biotinidase is the name of the enzyme that separates biotin from proteins and allows the body to reuse it.

Q. What happens to biotin in a child with partial biotinidase deficiency?

A. When there is not enough biotinidase enzyme or it does not work properly, biotin cannot be easily freed into the form that is used by the body. This results in low levels of biotin.

Q. What are the effects of having partial biotinidase deficiency if it is not treated?

A. A child with partial biotinidase deficiency may have some mild skin problems and hair loss. These can be prevented by treatment.

Q. What is the treatment for partial biotinidase deficiency?

A. The current recommendation is to treat partial biotinidase deficiency with daily supplements of biotin for six months. After that, treatment can be discontinued as there should be enough biotin in their normal diet.

Q. Why would a child have partial biotinidase deficiency?

A. Partial biotinidase deficiency is an inherited disorder. It results when a baby receives a biotinidase gene that does not work well from each parent. For more information about this, contact your health care provider or a genetic counselor.

Q. How common is partial biotinidase deficiency?

A. It is expected that about one in every 5,000 – 10,000 babies in Washington State will be born with partial biotinidase deficiency.

For more information about biotinidase deficiency, please see the Disorders section of our website:
www.doh.wa.gov/nbs.

